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Fibrous Dysplasia with Intramuscular Myxoma (Mazabraud's Syndrome).

*Report of a Case and Review of the Literature**

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ABSTRACT

About 30 cases of fibrous dysplasia associated with one or more myxomas (Mazabraud's syndrome) have been reported since 1926. We report a new case in a woman with polyostotic fibrous dysplasia and a myxoma in the left femoral muscle. She also had a history of precocious sexual development and café au lait spots, two manifestations whose association with polyostotic fibrous dysplasia defines McCune-Albright syndrome.

The first case of fibrous dysplasia and intramuscular myxoma was reported in 1926 by Henschen [1]. Mazabraud reported four additional cases and provided a detailed description of the condition in several articles published between 1957 and 1969 [2-6]. Only 30 cases or so have been reported to date. We report a new case in which the diagnosis of fibrous dysplasia was made during evaluation for a myxoma in the femoral muscle.

Case-report

A 37-year-old woman was evaluated for a steadily-growing tumor in the left thigh that she had first noticed one year earlier. Multiple areas of lysis and sclerosis were seen on radiographs of the femur (fig. 1) and were found to be hyperactive on a radionuclide bone scan. The patient was referred to our department.

A tangerine-sized, freely moveable, non-pulsating, painless tumor was felt in the anteromedial part of the left thigh. The temperature of the skin over the tumor was normal. Multiple café au lait spots were seen at various sites on the body. Other physical findings were unremarkable. The medical history revealed that menarche had occurred at 11 years of age.

Findings were normal from laboratory tests including markers for inflammation and assays of calcium and phosphate. An ultrasonogram of the thigh showed a sharply-defined hypoechoic mass with a few centrally-located fluid-filled cavities (fig. 2). Abnormalities strongly suggestive of fibrous dysplasia were seen on plain radiographs of the femurs and left tibia. A round, low-density, homogeneous mass measuring about 5 cm in diameter was visible on computed tomography scans of the left thigh. Magnetic resonance imaging demonstrated a well-defined mass generating a signal almost isointense to the surrounding muscle on T1 images, with postgadolinium enhancement. Very high signal from the mass was seen on T2 images. The mass was not in contact with the femur. These findings were consistent with an intramuscular fibroma arising from the femoral muscle (fig. 3). Sections through the femur showed signal abnormalities suggestive of fibrous dysplasia.

The tumor was removed surgically. It was separate from the bone and exhibited the gross appearance of a myxoid tumor.

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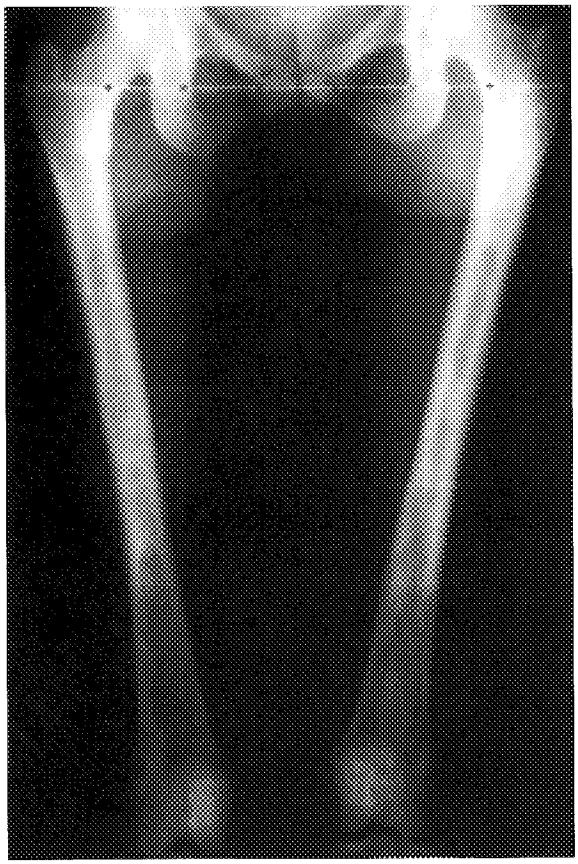

Fig. 1. - Radiograph of both femurs. Hypertrophic bone, mixed radiolucent and radioopaque lesions, cortical thinning (distal third) or hypertrophy, and disappearance of the bone structure.

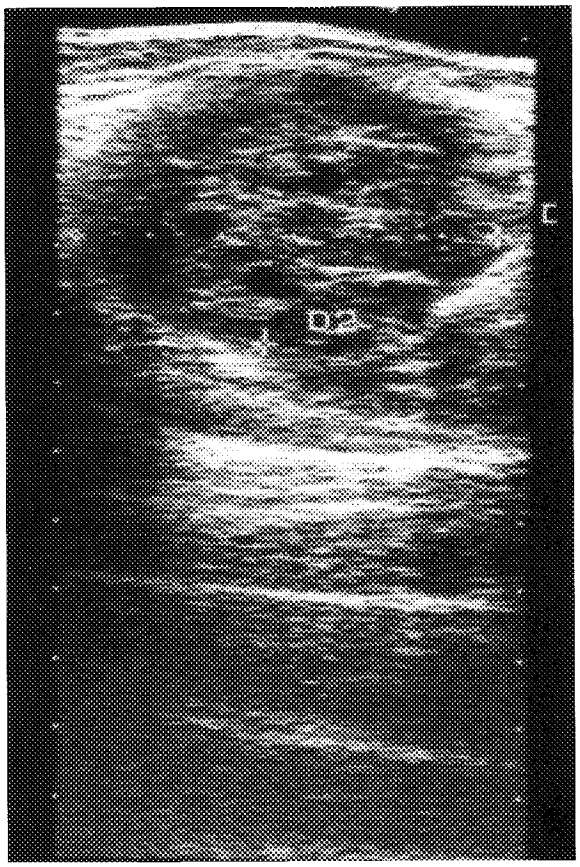

Fig. 2. - Ultrasonography of the left thigh. Sharply-demarcated hypoechoic mass containing several centrally-located fluid-filled cavities.

Histological findings established a diagnosis of intramuscular myxoma.

The diagnosis was Mazabraud's syndrome (fibrous dysplasia plus an intramuscular myxoma) with McCune-Albright syndrome (fibrous dysplasia plus precocious sexual development and café-au-lait spots).

Discussion

Fibrous dysplasia of bone is an infrequent, non-malignant condition of unknown cause. In a study of radiographs from 82 000 patients, 23 cases of fibrous dysplasia were identified [7]. The disease can be monostotic or polyostotic and is characterized by islands of fibrous metaplasia within flat and tubular bones. Polyostotic fibrous dysplasia is frequently unilateral in distribution. Most patients develop symptoms during childhood, consisting of a limp, bone deformities, bouts of pain, and pathological fractures. However, some cases are clinically silent. There is a well-established predominance in women over men.

Plain radiographs show evidence of bone dystrophy consisting of areas of hypertrophy, geodes bounded by fine sclerotic rims, a mixed radiolucent and radioopaque pattern, thin or hypertrophic cortices, or disappearance of the bone structure.

Laboratory test results are classically unremarkable. In particular, findings are normal from tests for inflammation and assays of calcium and phosphate in serum and urine. This, together with the radiographic findings, is usually sufficient to establish the diagnosis in polyostotic forms.

Fibrous dysplasia with endocrine disturbances (mainly precocious sexual development) and café au lait spots is known as McCune-Albright syndrome. Other endocrinopathies reported in this condition include goiter, hyperthyroidism with exophthalmos, and diabetes mellitus [8].

Only two cases of malignant transformation of the skeletal lesions have been reported in patients with Mazabraud's syndrome. Transformation to fibrosarcoma, osteosarcoma, or chondrosarcoma has been described on multiple occasions in patients with fibrous dysplasia but no myxoma, many of whom had a history of radiation therapy [9].

Myxoma is a benign, usually solitary tumor most common during the sixth and seventh decades of life.

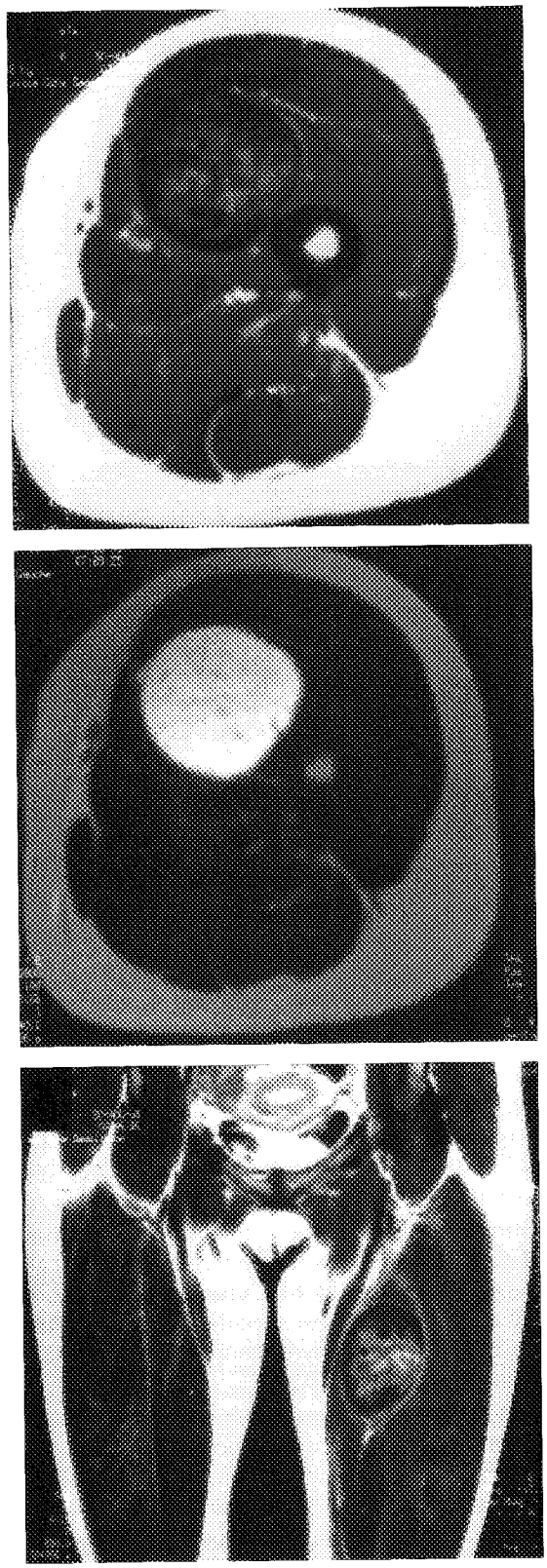

Fig. 3. - Magnetic resonance imaging of the left thigh. A: T1-weighted sequence after gadolinium injection, transversal section; B: T2-weighted sequence, transversal section. C: T1-weighted sequence after gadolinium injection, coronal section.

The main sites of involvement are the large muscles of the thighs, buttocks, and shoulders [10]. The myxomas seen in Mazabraud's syndrome are often multiple and arise in the vicinity of the skeletal lesions but remain separate from them. The clinical manifestations are those of myxomas in general, namely pain, development of a mass, or, exceptionally, signs of compression of a blood vessel or nerve. Recurrences are common after surgical removal [11].

Ultrasonography, a technique widely used to investigate soft tissue tumors, shows a sharply-defined hypoechoic mass containing a few centrally-located fluid-filled cavities. Computed tomography and magnetic resonance imaging are now routinely done as part of the preoperative workup and also show a well-defined tumor generating a hypointense or isointense signal on T1 images that enhances after gadolinium and a hyperintense signal on T2 images.

Several hypotheses have been put forward to explain the occurrence of myomas in Mazabraud's syndrome. In 1957, Mazabraud suggested that dysplasia of tendon-to-muscle junctions may be involved [2]. Similarities between myxomas and the synovial mucoid cysts produced by joint overloading have led to the suggestion that mechanical factors may promote the development of myxomas [12]. Two more cogent hypotheses involve metabolic abnormalities in soft tissues and bone during the growth period [13,14] and a gene mutation [15], respectively. None of these hypotheses has been confirmed to date.

The combination of cutaneous pigmentation, bone lesions, and subcutaneous tumors can in theory suggest other diagnoses such as neurofibromatosis or systemic mastocytosis. In practice, however, the appearance of the skin and bone lesions in Mazabraud's syndrome is distinctive, leaving little room for diagnostic error.

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